

N B D P S

NATIONAL BIRTH DEFECTS PREVENTION STUDY

NBDPS Update

The National Birth Defects Prevention Study (NBDPS) continues to grow and new cases are added daily. To date, the study sites have interviewed over 22,000 women. Also, over 9,000 families have sent in their cheek cell samples.

We are busy adding research topics. There are over 200 projects that will use data from the study. Research findings are being published. Two recent studies are described below. Within the next year, we plan to publish the results from 25 different studies. We thank all the families for allowing us to carry out the largest study ever on birth defects. This information will help us move closer to finding the causes of birth defects.

Selected Findings Using NBDPS Data

Allergy Drug and Hypospadias

Recently, a study from Sweden suggested that there might be a link between the allergy drug Claritin® and hypospadias. Hypospadias is a birth defect in boys where the urethral opening is on the underside of the penis instead of the tip. In the U.S., 7 of every 1000 boys are born with this birth defect.

Since Claritin® is a common allergy drug used in the U.S., it could affect the health of many families.

We used the data in this study to look at this issue. We studied 558 boys with severe hypospadias and 1432 boys with no birth defect. Each case had been reviewed by a doctor to make sure that it met our strict requirements for the study. We looked at the data to see whether the mother told us she had taken Claritin® one month before or in the first three months of her pregnancy. Our data did not show a higher risk of hypospadias among boys whose mothers took Claritin®. This should reassure women who took Claritin® before they knew they were pregnant. As always, women who are pregnant or planning a pregnancy should check with their doctors before taking any medications.

Reference: Centers for Disease Control and Prevention, *MMWR* 2004; 53(10): 219-221.
<http://www.cdc.gov/mmwr/preview/mmwrhtml/mm5310a5.htm>.

Muscular Ventricular Septal Defects and NSAIDs

One of the most common heart defects is a muscular ventricular septal defect (mVSD), an opening between two parts of the heart. A few studies have noted that there might be a higher risk of having a baby with an mVSD if a mom used nonsteroidal anti-inflammatory drugs (NSAIDs), or if she had a fever during her pregnancy. Some examples of NSAIDs are aspirin, ibuprofen, Aleve®, Advil®, and Motrin®.

-- continued on page 2

N E W S

2005

NBDPS News is a periodic newsletter of the Centers for Birth Defects Research and Prevention.

The purpose of this newsletter is to inform the public of the Centers' activities and current news about birth defects.



IN THIS ISSUE...

Page	Content
1NBDPS Update
2Smoking and Pregnancy
3NBDPS Interviewers
5AR CDBRP Director Named Endowed Chair in Birth Defects
5Unraveling the Mystery of Birth Defects Using Genomics
7Resources on Birth Defects And Related Topics
8Directory of CDBRP

Selected Findings Using NBDPS Data

-- continued from page 1

Using NBDPS data, we compared babies with an mVSD and babies without a birth defect. We looked at the data to see if the babies' mothers had taken any NSAIDs during her pregnancy. Our data did not show a higher risk for mVSD among babies whose mothers took these drugs. We also did not find a link between maternal fever and mVSD.

Reference: Cleves et al. Birth Defects Research (Part A) 2004; 70:107-113.

Smoking and Pregnancy

In the U.S., about 1 in 8 women smokes during pregnancy. Smoking may lower the oxygen available to the baby, which can cause the baby to grow slowly and gain less weight in the womb. But what do we know about smoking and birth defects?

Smoking Raises Risk for Oral Clefts

Oral clefts are birth defects that occur in the oral-facial region, often the lip, the roof of the mouth (hard palate), or the soft tissue in the back of the mouth (soft palate). Because of birth defects research, we now know that smokers' babies may be more likely to have cleft lip and/or cleft palate. One study looked at a gene that helps in the development of the palate and mouth. They found that:

- Women who smoked during pregnancy were almost twice as likely to have babies with oral clefts. The more cigarettes the mother smoked, the higher the risk.
- The hazards of smoking were even greater for the 1 in 7 babies who carry a cleft-susceptibility gene. Babies with this gene were 8 times as likely to have oral clefts *if their mothers smoked*. Those born to nonsmoking mothers with this gene were not more likely to have an oral cleft.



- Nonsmoking mothers who had been around second-hand smoke had only a small, if any, increased risk of having an oral cleft. But if both the mother and father smoked, there was a greater chance that the baby had an oral cleft.

Smoking and Other Birth Defects

- **Heart and limb defects.** In other birth defects studied, the connection with smoking is not as simple. For example, there was a somewhat higher risk for certain heart defects and limb defects, but only if both parents smoked. Perhaps smoking patterns are different when both parents smoke, or maybe there are other behaviors that are more common among smokers.
- **Neural tube defects.** Parents' smoking did not increase risk.
- **Down syndrome.** Babies with Down syndrome whose mothers smoked during the first trimester had twice the risk for heart defects compared to babies with Down syndrome whose mothers did not smoke during pregnancy.



What Can Women Do?

Pregnant women should avoid other people's smoke. Studies suggest that regular exposure to secondhand smoke may harm a developing child. If a woman smokes during pregnancy, she should quit or cut back as much as she can. Here are some resources that can help:

- A health care provider
- National Partnership to Help Pregnant Smokers Quit (<http://www.helppregnant smokersquit.org>)
- The American Legacy Foundation (<http://www.americanlegacy.org>)
- Smokefree.gov (<http://www.smokefree.gov>)

Sources: California Birth Defects Monitoring program (<http://www.cbddmp.org>) and March of Dimes Birth Defects Foundation (<http://www.marchofdimes.com>)

NBDPS Interviewers: A Face Behind the Voice

Most women who participate in this study come in contact with only one person—the interviewer who called on the phone and asked a lot of questions. Have you ever wondered what it's like to do their job?

We asked study interviewers to tell us what they enjoy about their work, what they find challenging, and what they would like to share with study participants. Samples of their responses are included here. We hope this gives you a better picture of the interviewers.

Before they make their first telephone call, interviewers must complete a lot of training. They learn how to:

- Follow study guidelines
- Speak the same way to each family
- Respect the rights of each family to refuse at any point, and
- Always maintain privacy

They are also very good with computers. It takes a lot of skill to move quickly through a computer-based form while doing an interview over the telephone.

Thoughts on my job as an NBDPS interviewer

“When I first considered the job of being a nurse-interviewer for a study on birth defects, I was hesitant. The job was about talking to women who had delivered babies with birth defects. It seemed as though I would be approaching these women at the absolute lowest points of their lives. How could I ask them to recount the events of the pregnancies? Wouldn't they be too caught up in their feelings of grief and guilt to want to answer questions about this time? I was sure that most women would refuse to participate in the study and would be angry with me for asking.

I couldn't have been more wrong. These women turned out to be an amazing group. Far from being at their worst, they are at their best. They have marshaled their efforts toward the challenge of caring for an infant with a birth defect. They are not only willing, but eager to participate in the study. Time after time, mothers tell me that they are glad to have a chance to help so that future moms may not have to go through what they have just experienced. I consider myself very lucky to have the chance to work with these women, and to be part of a nationwide effort to find out more about the causes of birth defects.” – *Massachusetts interviewer*

What do the interviewers enjoy most about their job? Why is this job important to them?

“As a project coordinator and main interviewer, there are many aspects of my job that I enjoy. However, I take joy in knowing the tasks of my job are vital. I note the importance of serving as a link between study participants and researchers. I collect information that could provide a breakthrough or create ways that could enhance the quality of life for a child. As a pediatric nurse, I was drawn to this job because it was a path that I had not traveled before and research has always been one of my interests.” – *Arkansas interviewer*

“What I like about my job is the contact with our mothers/respondents. We know that this is a chance for us to get the mother's story. In time this will help get us closer to understanding and preventing birth defects. Through this work, I have found out not only how common it is, but also how much families struggle and how painful it is.” – *California interviewer*

“I enjoy talking to moms and listening to their concerns. I guess that's a grandmother's instinct in me. What drew me to this job was that maybe I will be part of a large group helping to find ways to prevent birth defects. This job is important because I am learning some facts about what causes birth defects since one of my children might face some of these challenges.” – *Texas interviewer*

“Working as an NBDPS interviewer, I've been able to connect with so many amazing women who've opened their hearts to help others. I take pride in knowing that together with these strong mothers, I've been able to contribute to a study that can only bring knowledge, answers, and hope to families everywhere! It's important to me that I do the best job I can to make this study successful. We owe it to the mothers, babies, and families.” – *Interviewer for North Carolina and Atlanta CDC*

-- continued on page 4



Photos of NBDPS
Interviewers



Photos of NBDPS Interviewers

NBDPS Interviewers: A Face Behind the Voice

-- continued from page 3

“What drew me to this job is the potential it has to help future moms in having healthier babies. It is important because it makes me feel useful when I act as a link between the Hispanic community and the study.”

– *Spanish-language interviewer for North Carolina and Atlanta CDC*

What challenges do interviewers for the study face?

“What I do is important and as a part of my job, I’m collecting vital information that could enhance the lives of many families. As an interviewer I find that the challenge is that not everyone shares in the beliefs of the importance of research.” – *Arkansas interviewer*

“I have encountered many challenges in my job, but the first and foremost would be the emotional aspect. On a daily basis, I talk with mothers who have lost babies due to a number of reasons, and mothers who are taking care of babies who have birth defects. This can weigh heavily on a mother who has trouble adjusting with her loss or just coping with the child. Of course, these issues come through when being interviewed over the telephone. I find this to be the most difficult part of being an interviewer.” – *California interviewer*

“My greatest challenge is not being able to contact a mom.” – *Utah interviewer*

“One of my biggest challenges so far is working to fully understand and write down the most accurate answers each mom gives us. I want to record the information as accurately as possible from the moms for all the interview questions.”

– *Interviewer for Iowa and New York*

“I have to admit that doing these interviews can be emotionally draining, both for me and for the mothers. I hope they know we do feel for them, and that’s what makes this job worth every challenge.”

– *Interviewer for North Carolina and Atlanta CDC*

What would the interviewers want the moms to know about them or their job?

“I would like the moms that I interview to know that I’m a mom too. And there is never a need to apologize for family interruptions. I know that recalling information can be stressful.” – *Arkansas interviewer*

“I would like mothers to know that we do this work because we know it is important and we like doing it.” – *California interviewer*

“I would want mothers to know that I respect them a lot for everything they have been through. I want them to know that I appreciate every minute of their time spent talking to me. I want them to know that I’m doing this because I have hoped that with their cooperation we can push forward and change the future for thousands of children and their parents.”

– *Another California interviewer*



“Being a NBDPS interviewer is more than a job for me. It is a valuable learning experience. It is also a social contact in which I use many of the social skills that we all use in our every day lives such as politeness, friendliness, careful listening, sensitivity, etc. I really enjoy speaking with the moms and knowing that they are helping us understand more about birth defects, which will ultimately help prevent birth defects! I really enjoy the work I am doing, and I hope the data I am collecting helps to find the causes of birth defects.”

– *Interviewer for Iowa and New York*

“I would like for the moms to know that I am married. I don’t have any children, but I plan to have children in the future. I haven’t come across the experiences these women have had during pregnancy but I am grateful to hear them so I will be more prepared when the time comes for me.” – *Another Texas interviewer*

Arkansas Children's Hospital Awards the Pamela D. Stephens Endowed Chair in Birth Defects Research to Charlotte Hobbs, M.D., Ph.D.



Dr. Charlotte Hobbs was presented the Pamela D. Stephens Endowed Chair medallion by Dr. Debra Fiser, Chair, Department of Pediatrics and Dr. E. Albert Reece, Dean of the UAMS College of Medicine

Dr. Charlotte Hobbs was named the first holder of the Pamela D. Stephens Endowed Chair in Birth Defects Research. It recognizes Dr. Hobbs' work in the field of birth defects. This was formalized on August 12, 2004, at Arkansas Children's Hospital. Dr. Hobbs is the director of the Arkansas Center for Birth Defects Research and Prevention. She is also an Associate Professor and Section Chief of Birth Defects Research at Arkansas Children's Hospital and University of Arkansas for Medical Sciences. The Arkansas Children's Hospital is Arkansas's only children's hospital. It is also one of the ten largest children's hospitals in the nation.

Endowed chairs are vital in hiring and keeping the best and brightest. Being awarded an endowed chair is the highest honor paid to a researcher. The endowment fund was made possible by a gift made in memory of Pamela D. Stephens, who passed away in September 2003.

Unraveling the Mystery of Birth Defects Using Genomics

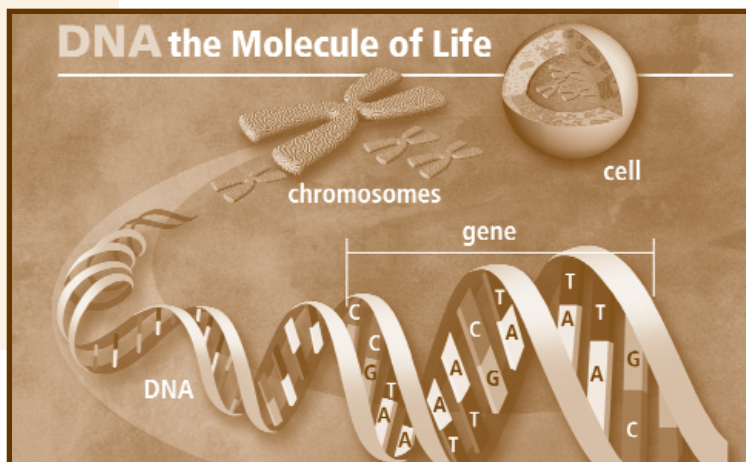
By Drs. Charlotte Hobbs and Patrycja Krakowiak,
Arkansas Center

The goal of the NBDPS is to provide insights into the causes of birth defects. Most birth defects result from a complex mix between genetic, environmental, and lifestyle factors. Women who take part in the NBDPS are first interviewed. Then the family is invited to give a sample of cheek cells from which their DNA is removed.

Some may wonder why getting DNA material from babies and their parents is such an important step in solving the mystery of birth defects. A key element of DNA is its chain of 4 building blocks, called nucleotides. Each is given a letter – A, T, C, and G. A only forms a bond with T, and G only forms a bond with C. Our genes act like a blueprint that lets our cells know how to make each of us a unique human being. Each person has about 30,000 genes on 23 pairs of chromosomes. About 99.9% of our DNA is the same from one person to another, but it is the 0.1% that varies between each person that makes us unique. Differences in DNA may make one baby more likely to have a birth defect than another baby. Finding out which nucleotides are a part of this process will be vital to developing future therapies for preventing birth defects.

The Human Genome project was completed in 2003. It created a blueprint of the entire length of the human DNA. Before it was done, scientists often did not know the areas of DNA that could prevent or cause common diseases. Now we have the opportunity to learn more about the causes of birth defects.

-- Continued on page 6



Source: U.S. Department of Energy Human Genome Program, <http://www.ornl.gov/hgmis>.

Unraveling the Mystery of Birth Defects Using Genomics

-- continued from page 5

Some people may be concerned about giving a sample of their DNA to NBDPS researchers. All of the information about the DNA is kept strictly private. Researchers studying the data use only the ID number and never see the names of the participants.

We have tried to make the process as simple as possible. The process to obtain DNA is painless and is almost like brushing the inside of a person's cheek with a toothbrush. Once we get the samples, anything that could name a person is removed and only a study ID number remains. Unique differences in the DNA are then compared between those with and without a birth defect.

Some participants have asked if the DNA can be used for cloning or for other reasons not listed on the consent forms. The answer is no. Although some animals have been cloned using DNA of other animals, it is not possible to use the DNA we get from our studies to clone another person.

Medical research studies during the last hundred years have helped to greatly improve the health of our babies and children. During this time, doctors and scientists



have found cures for polio, many childhood cancers, and infectious diseases. In 1915, 100 of every 1000 babies died before their first birthday. Due to major research findings, this number has fallen. Now, 7 of every 1000 babies die before their first birthday. In 1915, the major cause of infant death was infectious disease. In 2005, the leading cause of infant death is birth defects. NBDPS participants working with researchers can help to solve the mystery of birth defects by finding out what causes them. There are three major parts to the puzzle – environment, lifestyle, and genetics. By looking at the data from telephone interviews, we can find out what role the environment and lifestyle factors play. It is just as important to understand how our genes, our DNA, can add to the causes of birth defects.

The NBDPS is a national resource that can be used to find the risk factors for birth defects. When we understand the causes, then we can find ways to prevent birth defects.

At this point in history, we have a rare chance to begin to answer questions about the genetic causes of common birth defects and improve the health and quality of life for future babies. With DNA provided by families in this study, we will be able to find genetic factors that can lead to birth defects. Then we will be able to come up with useful prevention steps so that fewer babies will be born with birth defects. All of this is made possible because of the contributions of many families who have babies with and without birth defects.



Resources on Birth Defects and Related Topics

We have come up with a list of resources that might help you. The Centers are not responsible for the content found on these web sites.

Congenital Heart Defects

The **Children's Heart Institute** web site "helps parents understand the heart problems of their child." It has an online coloring book for kids to learn about the heart.
Web site: <http://childrenheartinstitute.org>

The **Heart Center Online** web site has useful information for heart patients and their health care providers. It has pictures and videos to show many aspects of the heart.
Main Web site: <http://www.heartcenteronline.com>
Animation (video) web site:
http://www.heartcenteronline.com/myheartdr/home/show_animations.cfm?cmbtopics=186

The **Congenital Heart Defects.com** web site has a list of information and resources on congenital heart disease.
Web site: <http://www.congenitalheartdefects.com>

The **American Heart Association** has information about heart disease, healthy living, and children's health. Some of the materials are available in Spanish.
Web site: <http://www.americanheart.org/presenter.jhtml?identifier=1200000>

PediHeart has heart disease information for both parents and children.
Web site: <http://www.pediheart.org/>

The **Congenital Heart Information Network** has information and resources about heart disease.
Web site:
<http://www.tchin.org/>



Cleft Lip/Palate

The **World Craniofacial Foundation** has information, resources, and support for children with head or face defects.
Address: 7777 Forest Lane, Ste C-621, P.O. Box 515838, Dallas, TX 75251-5838
Telephone: 1-800-533-3315
Web site: <http://worlddcf.org>

Operation Smile provides surgery for facial defects for qualified patients in the United States and throughout the world.
Address: 6435 Tidewater Drive, Norfolk, VA, 23509
Telephone: 1-888-677-6453
Web site: <http://operationsmile.org>

Support Groups

The **Family Village** has resources and information for persons with disabilities and their families.
Web site: <http://www.familyvillage.wisc.edu/index.htmlx>

The **Fathers' Network** web site supports fathers and their families who have a child with special health care needs.
Website: <http://www.fathersnetwork.org/>

Web sites Related to Genetics

The **Clinical Genetics Services at the University of Kansas Medical Center** has a web site called the Genetics and Rare Conditions Site. It lists the birth defects and genetic conditions in order from A to Z.
Web site: <http://www.kumc.edu/gec/support>

The **Mountain States Genetic Network** has information about national genetic support groups. This website also has materials in Spanish.
Web site: <http://www.mostgene.org/>

Web sites Related to Health

The **National Women's Health Information Center** has many topics on women's health. There are special sections on topics such as pregnancy and folic acid. This website also has materials in Spanish.
Web site: <http://www.4woman.gov>

Medline Plus has useful information such as health news, health topics, a medical dictionary, a medical encyclopedia, and drug information. Birth defects can be found under the list of health topics. The web site can be viewed in English or Spanish.
Web site: <http://medlineplus.gov>

DIRECTORY OF THE CENTERS FOR BIRTH DEFECTS RESEARCH AND PREVENTION

ARKANSAS

Charlotte Hobbs, MD, PhD
University of Arkansas for Medical Sciences
Arkansas Children's Hospital
Phone: (501) 364-5001; toll-free: (877) 662-4567
E-mail: hobbscharlotte@uams.edu
Internet: arbirthdefectsresearch.uams.edu

CALIFORNIA

Gary Shaw, DrPH
California Birth Defects Monitoring Program
Phone: (510) 549-4155
E-mail: gsh@cbdmp.org
Internet: www.cbdmp.org

GEORGIA/CDC

Margaret Honein, PhD, MPH
Centers for Disease Control and Prevention
Phone: (404) 498-4315
E-mail: nbdps@cdc.gov
Internet: www.cdc.gov/ncbddd

IOWA

Paul Romitti, PhD
University of Iowa
Phone: (866) 363-1980 (toll-free)
E-mail: paul-romitti@uiowa.edu
Internet: www.public-health.uiowa.edu/ircid

MASSACHUSETTS

Marlene Anderka, MPH
Massachusetts Department of Public Health
Phone: (617) 624-6045
E-mail: marlene.anderka@state.ma.us
Internet: www.mass.gov/birthdefectscenter

NEW YORK

Charlotte Druschel, MD, MPH
New York State Department of Health
Phone: (518) 402-7990; toll-free: (800) 458-1158, ext. 27990
E-mail: cmd05@health.state.ny.us
Internet: www.health.state.ny.us/nysdoh/cmr/cmrhome.htm

NORTH CAROLINA

Andy Olshan, PhD
University of North Carolina, Chapel Hill
School of Public Health
Phone: (877) 204-5994 (toll-free)
E-mail: andy_olshan@unc.edu
Internet: www.schs.state.nc.us/SCHS/bdmp

TEXAS

Mark Canfield, PhD
Texas Department of State Health Services
Phone: (512) 458-7232
E-mail: mark.canfield@dshs.state.tx.us
Internet: www.dshs.state.tx.us/birthdefects

UTAH

Marcia Feldkamp, PA, MSPH
Utah Department of Health
Phone: (866) 818-7096 (toll-free)
E-mail: mfeldkamp@utah.gov
Internet: www.health.utah.gov/birthdefect

If you no longer wish to receive this newsletter or would like to update your address, please let us know by calling toll-free at 1-800-814-1186 or emailing gpe@cbdmp.org. Please also let us know if you have topic ideas for future issues.

